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Neuroacanthocytosis Presenting with Psychiatric Symptoms

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ABSTRACT

An adult male presented with dyskinetic movements of the face and choreiform movements associated with dysphagia and vocaltics.Obsessive compulsive symptoms and depressive symptoms occurred 3 months after the onset of the illness.Peripheral blood smear revealed an excess of acanthocytes. A diagnosis of neuroacanthocytosis was made and he was started on treatment.

Key Words : Tics, obsessive compulsive symptoms, Neuroacanthocytosis

Introduction : Neuroacanthocytosis is a rare form of inherited choreoathetosis with an incidence of one in one lakh population. The characteristic features of this disorder are choreoathetosis,gait abnormalities,self injurious behaviour and axonal neuropathy (Bharucha et al .,1989). Psychiatric comorbidity in the form of depression, anxiety, personality change,cognitive impairments and obsessive compulsive symptoms have been described with neuroacanthocytosis (Hardie et al.,1991). The incidence of psychiatric symptoms in the early part of the illness is uncommon (Bruneau et al., 2003). Presence of tics and self injurious behaviour have been described in India in one patient(Galgali et al.,2001).We present a 45 year old man with a family history of choreoathetosis presenting with choreoathetosis,sensory symptoms and acanthocytes in the peripheral smear with onset of obsessive compulsive symptoms and depressive symptoms early in the illness. He is being reported to highlight the rarity of the condition and the incidence of psychiatric symptoms in the early part of the illness which is even rarer.

Case Report

A 45 year old male labourer from a low socio economic status Tamil speaking family presented with a progressive illness of one year duration characterized by a progressive dysphagia,dysarthria secondary to dyskinetic movements of the face,lips,tongue and the pharyngeal muscles.He also had choreiform movements involving the neck, shoulders, trunk and the lower limbs.There were also vocal tics in the form of clicking movements of the tongue.He developed symptoms of obsessive compulsive disorder in the form of obsessions of contamination and compulsions of washing and compulsive vocalization three months after the onset of abnormal movements. There were also depressive symptoms in the form of helplessness,hopelessness and suicidal ideation.He also reported impaired sensation in the lower limbs below the knee.There was no history to suggest a collagen vascular disease or an internal malignancy. There was no history to suggest a dyslipidemia or cognitive impairment. There was no history to suggest a motor or posterior column involvement. There were no other medical problems.There was history to suggest a similar illness in his elder brother who died of complications of the disease. There was no significant past or treatment history.

Systemic examination was normal except for flycatcher's tongue and dyskinetic movements of the uvula. Detailed central nervous system examination revealed choreoathetotic movements of the whole body. Higher mental functions,speech and cranial nerves were normal. There was no motor weakness with normal tone of muscles and normal reflexes.There were no sensory deficits. Mental status examination revealed obsessions of contamination and of self doubt associated with compulsive subvocalisation. Affect was depressed and non reactive, content of thought revealed depressive cognitions.

Investigations revealed a normal blood count.Peripheral smear showed acanthocytes in more than 90 percent of the field.Collagen workup was normal.There were no laboratory evidence of Wilson's disease.Fasting lipid profile was within normal limits.

Liver functions were normal.Electromyogram/nerve conduction velocity studies were normal.CT scan brain was

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normal.The diagnosis considered was neuroacanthocytosis on the basis of the above mentioned findings even though there was no self injurious behaviour. He was rated for obsessive compulsive symptoms using the YBOCS scale on which he scored 20. He scored 15 on the 17 item Hamilton depression rating scale for depressive symptoms. He was started on oral Haloperidol and Clonazapam for his dyskinetic movements with which he had improved marginally. Selective Serotonin Reuptake Inhibitors were also started for his obsessions and compulsions.

During serial follow ups,his choreiform movements had reduced along with his obsessive compulsive and depressive symptoms.He was seen once in 2 weeks for 3 months after the diagnosis was made.This patient will be regularly followed up in our out-patient clinic.

Neuroacanthocytosis otherwise called Levine Critchley syndrome is a rare form of inherited choreoathetosis associated with normal lipoproteins.It was first described by Levine in 1970.It is inherited as autosomal recessive with chromosomal locus at 9q21(Dobstone-Stone et al.,2000).The disorder is so named because of its association with acanthocytes in the peripheral smear.The mean age of onset is 32 years. The occurrence of psychiatric symptoms in the early part of the illness in uncommon and indicates a poor prognosis. There is no definitive treatment though trials of antioxidants to delay the progress of the disease are in progress.The treatment is mainly symptomatic and supportive (Sakai et al.,1985).

The mean life expectancy after diagnosis is about 10 years. Death is usually due to the self injurious behavior as a consequence of severe dyskinetic movements of the pharynx.

It is important for clinicians to routinely screen for acanthocytes in any patient presenting with abnormal involuntary movements associated with psychiatric symptoms.

A routine screen for psychiatric symptoms as listed above in every case of choreoathetosis along with assessment of cognitive functions is essential. It will also be necessary to rule out all other causes of choreoathetosis.

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